GREENVILLE MIDWIFERY CARE
PRENATAL GENETIC SCREENING QUESTIONNAIRE

Name: _____
MR#: _____

These questions apply to members in **both** your family and in the baby's father's family:

1. How old will you be when your baby is born? _____
2. How old will the father of the baby be when your baby is born? _____
3. Have you or the baby's father or anyone in either of your families ever had a baby or family member with:
   a. Down Syndrome or other chromosome abnormality? ☐ Yes ☐ No
   b. Spina Bifida or neural tube defect (open spine or brain)? ☐ Yes ☐ No
   c. Hydrocephalus (water on the brain)? ☐ Yes ☐ No
   d. Anencephaly (open or missing brain)? ☐ Yes ☐ No
   e. Hemophilia or other blood disorders (bleeds easily)? ☐ Yes ☐ No
   f. Muscular Dystrophy? ☐ Yes ☐ No
   g. Cystic Fibrosis? ☐ Yes ☐ No
   h. Congenital heart defect? ☐ Yes ☐ No
   i. Mental Retardation? ☐ Yes ☐ No
4. Have you or the baby's father or other close relative had:
   a. An infant born dead (stillborn)? ☐ Yes ☐ No
   b. Three or more pregnancy losses? ☐ Yes ☐ No
   c. A baby with a birth defect? ☐ Yes ☐ No
5. Are you or the baby's father of Mediterranean descent, or have a history of Thalassemia? ☐ Yes ☐ No
6. Do you or the baby's father have sickle cell trait or anemia? ☐ Yes ☐ No
7. Do you or the baby's father have Jewish relatives, or have a family history of Tay-Sachs? ☐ Yes ☐ No
8. Do you know of any genetic (inherited) health problem in your family or the father's family? ☐ Yes ☐ No
9. Is there any other health problem that you are concerned about in you or your baby's father? ☐ Yes ☐ No
   Explain: _____

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- **Nuchal Translucency Ultrasound** is a screening test for Down syndrome or other abnormalities of chromosomal development. The most common are Down syndrome and trisomy 13 and 18. Down syndrome occurs at a rate of 1 in 700 babies. Trisomy 13 occurs at a rate of 1 in 10,000 and trisomy 18 occurs at a rate of 1 in 8000. First trimester screening will detect about 95% of pregnancies with trisomy 13 or 18.

  A combination blood test and ultrasound are performed between 11-14 weeks of pregnancy. This ultrasound measures the thickness at the back of the baby's neck. All babies have fluid at the back of their neck, but in Down syndrome or other abnormalities there may be more fluid than normal. The blood tests measure the amounts of human chorionic gonadotropin (hCG) and pregnancy-associated plasma protein (PAPP-A) which may be decreased in babies with Down Syndrome.

  I want NT Screening _____ ☐ Yes ☐ No

- **MaterniT21:** A blood test for women at increased risk of certain birth defects. This test detects increased amounts of material from chromosome 21 (Down Syndrome), chromosome 13 (Trisomy 13) and chromosome 18 (Trisomy 18). This test has high detection rates and low false-positive rates for these abnormalities:

    - 99.1% detection for Down Syndrome
    - 99.1% detection for trisomy 18
    - 91.7% detection for trisomy 13

- **QUAD screen**: A screening blood test performed between 16- 20 weeks of pregnancy. It is a screening for Down syndrome and open-spine defects, often referred to as "open neural tube defects" (ONTD). These defects occur at a rate of 1-4 per 1000 pregnancies. This test measures levels of 4 specific substances in the mother's blood:

    - AFP: alpha-fetoprotein, a protein produced by the fetus
    - hCG: hormone produced by the placenta
    - estriol: an estrogen produced by the fetus and the placenta
    - inhibin-A: protein produced by the placenta and ovaries

    The results are combined with the mother's age and ethnicity in order to determine the risk of potential disorders.

    I want Multiple Marker screening _____ ☐ Yes ☐ No

- **Cystic Fibrosis** : Cystic fibrosis is a life-threatening disease that primarily affects the lungs and digestive system. Some people are carriers of the gene even though they do not have the disease. There are currently no treatments available during pregnancy if your baby has cystic fibrosis, but it can be managed and controlled with specialized care through infancy through adulthood. A blood test is available to determine whether or not you are a carrier for most of the genes that cause cystic fibrosis. However, not all cystic fibrosis is detectable with testing.

    I want Cystic Fibrosis Carrier Screening _____ ☐ Yes ☐ No

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I understand that genetic counseling and prenatal diagnosis can be arranged if I desire.

_____

Patient Signature Date

_____

Provider or nurse signature Date

Genetic Screening Referral: ☐ No ☐ Yes ☐ Declined

Appointment Date / Time: _____

_____

Reviewed By Date

JFPCNM Aug 2014